OP-120 A RARE COMPLICATION AFTER TOTAL EXCISION OF FRONTAL AVM IN THE PEDIATRIC ERA: LOSS OF VISION IN ONE EYE

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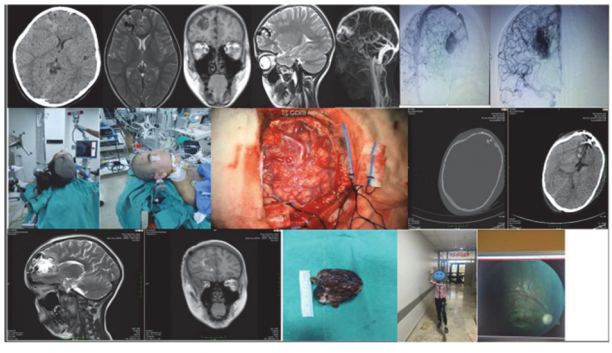
Aim Brain arteriovenous malformations (AVM)are defined as high-flow congenital masses consisting of nidus and drainage veins that may be of different sizes from the feeding arteries. They rarely present in childhood. Childhood AVM constitute 3–20%of all AVM. Although it is rare in childhood, 30–50% of intracranial hemorrhages in this age are caused by AVM. We aimed to present our case, which presented with complaints of headache, seizures and tendency to sleep, and resulted in total vision loss in the right eye as a result of central retinal artery stenosis that developed in the early period after longterm surgery.

Material and Method A 13-year-old patient was admitted to our hospital with complaints of sudden seizures and confusion and a referral request from an external center. The patient's GCS:14, she was prone to sleep. The patient, who had no motor deficit, had incontinence during the seizure. Other neurological examination findings were normal.

Results CT-CT angiography and DSA angiography were performed on the patient. DSA angiography;It was reported as a frontal AVM with feeders from 4 places (A2, MCA)and 2 drainage veins. At the neuroradiology council, it was decided to undergo surgery (figure 1).

Conclusions The frontal AVM was completely excised after finding and clipping the feeders and drainage veins one by one with a microsurgery method that took approximately 20 hours. It was stated that the patient, who had no motor sensory deficit in the postoperative period, lost vision in the right eye on the 3rd day. As a result of eye consultation, occlusion was detected in the right retinal artery. It was recommended that the patient be given urgent hyperbaric O2 therapy. Patient, who received 10 sessions of hyperbaric O2 treatment, stated that vision loss in the right eye continued. To our knowledge, no cases of total retinal artery occlusion afterAVM surgery have been reported in the literature. It is due to vasospasm or microembolism in prolonged surgery is considered. Even if slightly successful results are obtained with hyperbaric O2 therapy, its treatment still remains a problem.



Abstract OP-120 Figure 1 Preoperative, intraoperative and postoperative images (figure 1). Preop Ct, MRI, DSA-Intraop Figure-Postop CT, MRI, Eye Anjiography.

OP-121 DIAGNOSTIC YIELD OF WES IN PEDIATRIC PATIENTS: A SINGLE-CENTER EXPERIENCE

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Aim This study aims to assess the diagnostic yield of Whole Exome Sequencing (WES) in pediatric patients presenting with diverse clinical indications within a specialized genetic evaluation center.

Material and Method WES was conducted on 741 pediatric patients, presenting with neurological, metabolic, musculoskeletal, cardiovascular, sensory, autoimmune, and complex conditions. Approximately 20% of patients originated from consanguineous families. Exonic regions were sequenced at an average coverage of 100X using the Novaseq 6000 next-generation sequencing platform. Bioinformatic analysis followed the manufacturer's recommended workflow, and variant analysis adhered to ACMG criteria, ClinVar, and HGMD. Sanger sequencing with region-specific primers was employed for segregation analysis.

Results WES analysis identified reportable pathogenic variants and/or variants of uncertain significance (VUS) in 548 out of 741 patients with neurological (73.5%, n=511), metabolic (71.4%, n=42), musco-skelatal (78%, n=41), autoimmune (58.8%, n=34), skin (83.3%, n=6), cardiovascular (72.7%, n=11), sensory (100%, n=7) and multiple conditions (77.7%, n=63). Segregation analysis in 235 families confirmed the diagnosis in 181 cases, resulting in an overall diagnostic yield of 24.4% (181/741). Notably, 29 families with confirmed diagnoses underwent in vitro fertilization (IVF) and preimplantation genetic testing for monogenic disorders (PGT-M), leading to the successful birth of healthy infants.

Conclusions Following the exclusion of patients with only Variants of Uncertain Significance (VUS) and the inclusion of segregation analysis, the overall diagnostic confirmation rate reached 24.2%. To further improve confirmations, an extension of segregation analyses to include previously untested families (n=313) is warranted. Nevertheless, an acceptable diagnostic yield achieved in this study is notably influenced by the substantial prevalence of consanguineous cases. These findings underscore the crucial role of WES in pediatric conditions, not only in identifying novel and/or actionable genes with therapeutic applications but also in employing additional technologies, such as IVF and PGT-M, to prevent rare genetic disorders in subsequent generations within affected families.